IL2RG gene

interleukin 2 receptor subunit gamma

Normal Function

The *IL2RG* gene provides instructions for making a protein called the common gamma chain. This protein is a component of several different receptors that are involved in immune system function. The receptors span the cell membrane, with one end outside the cell like an antenna and the other end inside to transmit signals to the nucleus. Other proteins attach to these receptors, like a key in a lock, to trigger a series of chemical reactions inside the cell.

Receptors containing the common gamma chain are located on the surface of immature blood-forming cells in bone marrow. They partner with other proteins to direct blood-forming cells to form lymphocytes (a type of white blood cell). The receptors also regulate the growth and maturation of several subtypes of lymphocytes: T cells, B cells, and natural killer cells. These cells kill viruses, make antibodies, and help regulate the entire immune system.

Health Conditions Related to Genetic Changes

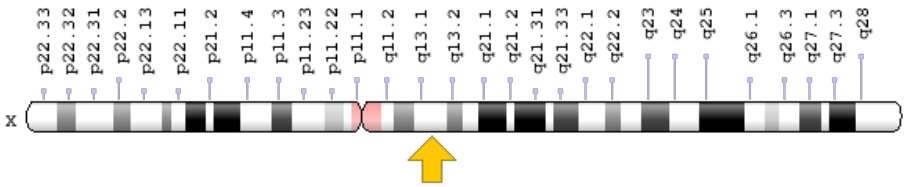
X-linked severe combined immunodeficiency

More than 300 mutations in the *IL2RG* gene have been identified in people with X-linked severe combined immunodeficiency (SCID). Most of these mutations involve changes in one or a few DNA building blocks (nucleotides) in the gene. These changes lead to the production of a nonfunctional version of the common gamma chain or prevent any protein from being produced. Without the common gamma chain, important chemical signals are not relayed to the nucleus and lymphocytes cannot develop normally. A lack of functional mature lymphocytes prevents the immune system from fighting off infections.

Chromosomal Location

Cytogenetic Location: Xq13.1, which is the long (q) arm of the X chromosome at position 13.1

Molecular Location: base pairs 71,107,404 to 71,111,631 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CD132
- common cytokine receptor gamma chain
- Gamma-C
- IL2RG_HUMAN
- IMD4
- interleukin 2 receptor, gamma
- interleukin 2 receptor, gamma (severe combined immunodeficiency)
- SCIDX
- SCIDX1
- γc

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): Defects in T-cell function result in severe combined immunodeficiencies
  https://www.ncbi.nlm.nih.gov/books/NBK27109/#A1509

GeneReviews

- X-Linked Severe Combined Immunodeficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1410

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28IL2RG%5BTIAB%5D%29+OR+%28%28CD132%5BTIAB%5D%29+OR+%28common+cytokine+receptor+gamma+chain%5BTI%5D%29+OR+%28IMD4%5BTIAB%5D%29+OR+%28SCIDX%5BTIAB%5D%29+OR+%28common+gamma+chain%5BTI%5D%29+OR+%28IL-2+receptor%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- INTERLEUKIN 2 RECEPTOR, GAMMA
  http://omim.org/entry/308380

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_IL2RG.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=IL2RG%5Bgene%5D
- HGNC Gene Family: CD molecules
  http://www.genenames.org/cgi-bin/genefamilies/set/471
- HGNC Gene Family: Fibronectin type III domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/555
- HGNC Gene Family: Interleukin receptors
  http://www.genenames.org/cgi-bin/genefamilies/set/602
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6010
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3561
- UniProt
  http://www.uniprot.org/uniprot/P31785

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15032591
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